

PAX3

References

Sort

- Medic, S., Rizos, H. and Ziman, M. (2011). Differential PAX3 functions in normal skin melanocytes and melanoma cells. *Biochemical and biophysical research communications*, 411, 832-7. [\[PubMed\]](#)
- Betters, E., Liu, Y., Kjaeldgaard, A., Sundstrom, E. and Garcia-Castro, M. (2010). Analysis of early human neural crest development. *Dev Biol.* [\[PubMed\]](#)
- Degenhardt, K. R., Milewski, R. C., Padmanabhan, A., Miller, M., Singh, M. K., Lang, D., Engleka, K. A., Wu, M., Li, J., Zhou, D., et al. (2010). Distinct enhancers at the Pax3 locus can function redundantly to regulate neural tube and neural crest expressions. *Developmental biology*, 339, 519-27. [\[PubMed\]](#)
- Guo, X. L., Ruan, H. B., Li, Y., Gao, X. and Li, W. (2010). Identification of a novel nonsense mutation on the Pax3 gene in ENU-derived white belly spotting mice and its genetic interaction with c-Kit. *Pigment Cell Melanoma Res*, 23, 252-62. [\[PubMed\]](#)
- He, S., Yoon, H. S., Suh, B. J. and Eccles, M. R. (2010). PAX3 Is extensively expressed in benign and malignant tissues of the melanocytic lineage in humans. *J Invest Dermatol*, 130, 1465-8. [\[PubMed\]](#)
- Lai, I. L., Lin, T. P., Yao, Y. L., Lin, C. Y., Hsieh, M. J. and Yang, W. M. (2010). Histone deacetylase 10 relieves repression on the melanogenic program by maintaining the deacetylation status of repressors. *J Biol Chem*, 285, 7187-96. [\[PubMed\]](#)
- Mascarenhas, J. B., Littlejohn, E. L., Wolsky, R. J., Young, K. P., Nelson, M., Salgia, R. and Lang, D. (2010). PAX3 and SOX10 activate MET receptor expression in melanoma. *Pigment Cell Melanoma Res*, 23, 225-37. [\[PubMed\]](#)
- Medic, S. and Ziman, M. (2010). PAX3 expression in normal skin melanocytes and melanocytic lesions (naevi and melanomas). *PLoS One*, 5, e9977. [\[PubMed\]](#)
- Birrane, G., Soni, A. and Ladis, J. A. (2009). Structural Basis for DNA Recognition by the Human PAX3 Homeodomain (dagger) (,) (double dagger). *Biochemistry*. [\[PubMed\]](#)
- Hilari, J. M., Mangas, C., Xi, L., Paradelo, C., Ferrandiz, C., Hughes, S. J., Yueh, C., Altomare, I., Gooding, W. E. and Godfrey, T. E. (2009). Molecular staging of pathologically negative sentinel lymph nodes from melanoma patients using multimarker, quantitative real-time rt-PCR. *Ann Surg Oncol*, 16, 177-85. [\[PubMed\]](#)
- Nakazaki, H., Shen, Y. W., Yun, B., Reddy, A., Khanna, V., Mania-Farnell, B., Ichi, S., Mcclone, D. G., Tomita, T. and Mayanil, C. S. (2009). Transcriptional regulation by Pax3 and TGFbeta2 signaling: a potential gene regulatory network in neural crest development. *Int J Dev Biol*, 53, 69-79. [\[PubMed\]](#)
- Thomas, A. J. and Erickson, C. A. (2009). FOXD3 regulates the lineage switch between neural crest-derived glial cells and pigment cells by repressing MITF through a non-canonical mechanism. *Development*, 136, 1849-58. [\[PubMed\]](#)
- Corry, G. N., Hendzel, M. J. and Underhill, D. A. (2008). Subnuclear localization and mobility are key indicators of PAX3 dysfunction in Waardenburg syndrome. *Hum Mol Genet*, 17, 1825-37. [\[PubMed\]](#)

Fenby, B. T., Fotaki, V. and Mason, J. O. (2008). Pax3 regulates Wnt1 expression via a conserved binding site in the 5' proximal promoter. *Biochim Biophys Acta*, 1779, 115-21. [\[PubMed\]](#)

Minchin, J. E. and Hughes, S. M. (2008). Sequential actions of Pax3 and Pax7 drive xanthophore development in zebrafish neural crest. *Dev Biol*, 317, 508-22. [\[PubMed\]](#)

Nakazaki, H., Reddy, A. C., Mania-Farnell, B. L., Shen, Y. W., Ichi, S., Mccabe, C., George, D., Mcclone, D. G., Tomita, T. and Mayanil, C. S. (2008). Key basic helix-loop-helix transcription factor genes Hes1 and Ngn2 are regulated by Pax3 during mouse embryonic development. *Dev Biol*, 316, 510-23. [\[PubMed\]](#)

Plummer, R. S., Shea, C. R., Nelson, M., Powell, S. K., Freeman, D. M., Dan, C. P. and Lang, D. (2008). PAX3 expression in primary melanomas and nevi. *Mod Pathol*, 21, 525-30. [\[PubMed\]](#)

Stoller, J. Z., Degenhardt, K. R., Huang, L., Zhou, D. D., Lu, M. M. and Epstein, J. A. (2008). Cre reporter mouse expressing a nuclear localized fusion of GFP and beta-galactosidase reveals new derivatives of Pax3-expressing precursors. *Genesis*, 46, 200-4. [\[PubMed\]](#)

Wu, M., Li, J., Engleka, K. A., Zhou, B., Lu, M. M., Plotkin, J. B. and Epstein, J. A. (2008). Persistent expression of Pax3 in the neural crest causes cleft palate and defective osteogenesis in mice. *J Clin Invest*, 118, 2076-87. [\[PubMed\]](#)

Yang, G., Li, Y., Nishimura, E. K., Xin, H., Zhou, A., Guo, Y., Dong, L., Denning, M. F., Nickoloff, B. J. and Cui, R. (2008). Inhibition of PAX3 by TGF-beta modulates melanocyte viability. *Mol Cell*, 32, 554-63. [\[PubMed\]](#)

Zhou, H. M., Wang, J., Rogers, R. and Conway, S. J. (2008). Lineage-specific responses to reduced embryonic Pax3 expression levels. *Dev Biol*, 315, 369-82. [\[PubMed\]](#)

Apuzzo, S. and Gros, P. (2007). Cooperative interactions between the two DNA binding domains of Pax3: helix 2 of the paired domain is in the proximity of the amino terminus of the homeodomain. *Biochemistry*, 46, 2984-93. [\[PubMed\]](#)

Lacosta, A. M., Canudas, J., Gonzalez, C., Muniesa, P., Sarasa, M. and Dominguez, L. (2007). Pax7 identifies neural crest, chromatophore lineages and pigment stem cells during zebrafish development. *Int J Dev Biol*, 51, 327-31. [\[PubMed\]](#)

Li, H. G., Wang, Q., Li, H. M., Kumar, S., Parker, C., Slevin, M. and Kumar, P. (2007). PAX3 and PAX3-FKHR promote rhabdomyosarcoma cell survival through downregulation of PTEN. *Cancer Lett*, 253, 215-23. [\[PubMed\]](#)

Milunsky, J. M., Maher, T. A., Ito, M. and Milunsky, A. (2007). The value of MLPA in Waardenburg syndrome. *Genet Test*, 11, 179-82. [\[PubMed\]](#)

Ryu, B., Kim, D. S., Deluca, A. M. and Alani, R. M. (2007). Comprehensive expression profiling of tumor cell lines identifies molecular signatures of melanoma progression. *PLoS ONE*, 2, e594. [\[PubMed\]](#)

Underwood, T. J., Amin, J., Lillycrop, K. A. and Blaydes, J. P. (2007). Dissection of the functional interaction between p53 and the embryonic proto-oncoprotein PAX3. *FEBS Lett*, 581, 5831-5. [\[PubMed\]](#)

Wang, Q., Kumar, S., Mitsios, N., Slevin, M. and Kumar, P. (2007). Investigation of downstream target genes of PAX3c, PAX3e and PAX3g isoforms in melanocytes by microarray analysis. *Int J Cancer*, 120, 1223-31. [\[PubMed\]](#)

Bajard, L., Relaix, F., Lagha, M., Rocancourt, D., Daubas, P. and Buckingham, M. E. (2006). A novel genetic

hierarchy functions during hypaxial myogenesis: Pax3 directly activates Myf5 in muscle progenitor cells in the limb. *Genes Dev*, 20, 2450-64. [\[PubMed\]](#)

Hsieh, M. J., Yao, Y. L., Lai, I. L. and Yang, W. M. (2006). Transcriptional repression activity of PAX3 is modulated by competition between corepressor KAP1 and heterochromatin protein 1. *Biochem Biophys Res Commun*, 349, 573-81. [\[PubMed\]](#)

Mayanil, C. S., Pool, A., Nakazaki, H., Reddy, A. C., Mania-Farnell, B., Yun, B., George, D., Mcclone, D. G. and Bremer, E. G. (2006). Regulation of murine TGFbeta2 by Pax3 during early embryonic development. *J Biol Chem*, 281, 24544-52. [\[PubMed\]](#)

Murakami, M., Tominaga, J., Makita, R., Uchijima, Y., Kurihara, Y., Nakagawa, O., Asano, T. and Kurihara, H. (2006). Transcriptional activity of Pax3 is co-activated by TAZ. *Biochem Biophys Res Commun*, 339, 533-9. [\[PubMed\]](#)

Rodeberg, D. A., Nuss, R. A., Elsawa, S. F., Erskine, C. L. and Celis, E. (2006). Generation of tumoricidal PAX3 peptide antigen specific cytotoxic T lymphocytes. *Int J Cancer*, 119, 126-32. [\[PubMed\]](#)

Tshori, S., Gilon, D., Beeri, R., Nechushtan, H., Kaluzhny, D., Pikarsky, E. and Razin, E. (2006). Transcription factor MITF regulates cardiac growth and hypertrophy. *J Clin Invest*, 116, 2673-81. [\[PubMed\]](#)

Wang, Q., Kumar, S., Slevin, M. and Kumar, P. (2006). Functional analysis of alternative isoforms of the transcription factor PAX3 in melanocytes in vitro. *Cancer Res*, 66, 8574-80. [\[PubMed\]](#)

Blake, J. A. and Ziman, M. R. (2005). Pax3 transcripts in melanoblast development. *Dev Growth Differ*, 47, 627-35. [\[PubMed\]](#)

Brown, C. B., Engleka, K. A., Wenning, J., Min Lu, M. and Epstein, J. A. (2005). Identification of a hypaxial somite enhancer element regulating Pax3 expression in migrating myoblasts and characterization of hypaxial muscle Cre transgenic mice. *Genesis*, 41, 202-9. [\[PubMed\]](#)

Corry, G. N. and Underhill, D. A. (2005). Pax3 target gene recognition occurs through distinct modes that are differentially affected by disease-associated mutations. *Pigment Cell Res*, 18, 427-38. [\[PubMed\]](#)

Engleka, K. A., Gitler, A. D., Zhang, M., Zhou, D. D., High, F. A. and Epstein, J. A. (2005). Insertion of Cre into the Pax3 locus creates a new allele of Splotch and identifies unexpected Pax3 derivatives. *Dev Biol*, 280, 396-406. [\[PubMed\]](#)

Gershon, T. R., Oppenheimer, O., Chin, S. S. and Gerald, W. L. (2005). Temporally regulated neural crest transcription factors distinguish neuroectodermal tumors of varying malignancy and differentiation. *Neoplasia*, 7, 575-84. [\[PubMed\]](#)

He, S. J., Stevens, G., Braithwaite, A. W. and Eccles, M. R. (2005). Transfection of melanoma cells with antisense PAX3 oligonucleotides additively complements cisplatin-induced cytotoxicity. *Mol Cancer Ther*, 4, 996-1003. [\[PubMed\]](#)

Koyanagi, K., Kuo, C., Nakagawa, T., Mori, T., Ueno, H., Lorico, A. R., Jr., Wang, H. J., Hseuh, E., O'day, S. J. and Hoon, D. S. (2005a). Multimarker quantitative real-time PCR detection of circulating melanoma cells in peripheral blood: relation to disease stage in melanoma patients. *Clin Chem*, 51, 981-8. [\[PubMed\]](#)

Koyanagi, K., O'day, S. J., Gonzalez, R., Lewis, K., Robinson, W. A., Amatruda, T. T., Wang, H. J., Elashoff, R. M., Takeuchi, H., Umetani, N., et al. (2005b). Serial monitoring of circulating melanoma cells during neoadjuvant biochemotherapy for stage III melanoma: outcome prediction in a multicenter trial. *J Clin Oncol*, 23, 8057-64. [\[PubMed\]](#)

Lacosta, A. M., Muniesa, P., Ruberte, J., Sarasa, M. and Dominguez, L. (2005). Novel expression patterns of Pax3/Pax7 in early trunk neural crest and its melanocyte and non-melanocyte lineages in amniote embryos. *Pigment Cell Res*, 18, 243-51. [\[PubMed\]](#)

Lang, D., Lu, M. M., Huang, L., Engleka, K. A., Zhang, M., Chu, E. Y., Lipner, S., Skoultchi, A., Millar, S. E. and Epstein, J. A. (2005). Pax3 functions at a nodal point in melanocyte stem cell differentiation. *Nature*, 433, 884-7. [\[PubMed\]](#)

Matsuzaki, Y., Hashimoto, S., Fujita, T., Suzuki, T., Sakurai, T., Matsushima, K. and Kawakami, Y. (2005). Systematic identification of human melanoma antigens using serial analysis of gene expression (SAGE). *J Immunother*, 28, 10-9. [\[PubMed\]](#)

Relaix, F., Rocancourt, D., Mansouri, A. and Buckingham, M. (2005). A Pax3/Pax7-dependent population of skeletal muscle progenitor cells. *Nature*, 435, 948-53. [\[PubMed\]](#)

Tatzel, J., Poser, I., Schroeder, J. and Bosserhoff, A. K. (2005). Inhibition of melanoma inhibitory activity (MIA) expression in melanoma cells leads to molecular and phenotypic changes. *Pigment Cell Res*, 18, 92-101. [\[PubMed\]](#)

Zhu, B. K. and Pruitt, S. C. (2005). Determination of transcription factors and their possible roles in the regulation of Pax3 gene expression in the mouse B16 F1 melanoma cell line. *Melanoma Res*, 15, 363-73. [\[PubMed\]](#)

Apuzzo, S., Abdelhakim, A., Fortin, A. S. and Gros, P. (2004). Cross-talk between the paired domain and the homeodomain of Pax3: DNA binding by each domain causes a structural change in the other domain, supporting interdependence for DNA Binding. *J Biol Chem*, 279, 33601-12. [\[PubMed\]](#)

Bogani, D., Warr, N., Elms, P., Davies, J., Tymowska-Lalanne, Z., Goldsworthy, M., Cox, R. D., Keays, D. A., Flint, J., Wilson, V., et al. (2004). New semidominant mutations that affect mouse development. *Genesis*, 40, 109-117. [\[PubMed\]](#)

Hou, L., Loftus, S. K., Incao, A., Chen, A. and Pavan, W. J. (2004). Complementation of melanocyte development in SOX10 mutant neural crest using lineage-directed gene transfer. *Dev Dyn*, 229, 54-62. [\[PubMed\]](#)

Kamaraju, A. K., Adjalley, S., Zhang, P., Chebath, J. and Revel, M. (2004). C/EBP-delta induction by gp130 signaling. Role in transition to myelin gene expressing phenotype in a melanoma cell line model. *J Biol Chem*, 279, 3852-61. [\[PubMed\]](#)

Keller, C., Arenkiel, B. R., Coffin, C. M., El-Bardeesy, N., Depinho, R. A. and Capecchi, M. R. (2004). Alveolar rhabdomyosarcomas in conditional Pax3:Fkhr mice: cooperativity of Ink4a/ARF and Trp53 loss of function. *Genes Dev*, 18, 2614-26. [\[PubMed\]](#)

Milewski, R. C., Chi, N. C., Li, J., Brown, C., Lu, M. M. and Epstein, J. A. (2004). Identification of minimal enhancer elements sufficient for Pax3 expression in neural crest and implication of Tead2 as a regulator of Pax3. *Development*, 131, 829-37. [\[PubMed\]](#)

Parker, C. J., Shawcross, S. G., Li, H., Wang, Q. Y., Herrington, C. S., Kumar, S., Mackie, R. M., Prime, W., Rennie, I. G., Sisley, K., et al. (2004). Expression of PAX 3 alternatively spliced transcripts and identification of two new isoforms in human tumors of neural crest origin. *Int J Cancer*, 108, 314-20. [\[PubMed\]](#)

Ploski, J. E., Shamsher, M. K. and Radu, A. (2004). Paired-type homeodomain transcription factors are imported into the nucleus by karyopherin 13. *Mol Cell Biol*, 24, 4824-34. [\[PubMed\]](#)

Pruitt, S. C., Bussman, A., Maslov, A. Y., Natoli, T. A. and Heinaman, R. (2004). Hox/Pbx and Brn binding sites mediate Pax3 expression in vitro and in vivo. *Gene Expr Patterns*, 4, 671-85. [\[PubMed\]](#)

Relaix, F., Rocancourt, D., Mansouri, A. and Buckingham, M. (2004). Divergent functions of murine Pax3 and Pax7 in limb muscle development. *Genes Dev*, 18, 1088-105. [\[PubMed\]](#)

Takeuchi, H., Morton, D. L., Kuo, C., Turner, R. R., Elashoff, D., Elashoff, R., Taback, B., Fujimoto, A. and Hoon, D. S. (2004). Prognostic significance of molecular upstaging of paraffin-embedded sentinel lymph nodes in melanoma patients. *J Clin Oncol*, 22, 2671-80. [\[PubMed\]](#)

Sommer, A. and Bartholomew, D. W. (2003). Craniofacial-deafness-hand syndrome revisited. *Am J Med Genet A*, 123A, 91-4. [\[PubMed\]](#)

Chang, T. I., Horal, M., Jain, S. K., Wang, F., Patel, R. and Loeken, M. R. (2003). Oxidant regulation of gene expression and neural tube development: Insights gained from diabetic pregnancy on molecular causes of neural tube defects. *Diabetologia*, 46, 538-45. [\[PubMed\]](#)

Lang, D. and Epstein, J. A. (2003). Sox10 and Pax3 physically interact to mediate activation of a conserved c-RET enhancer. *Hum Mol Genet*, 12, 937-45. [\[PubMed\]](#)

Muratovska, A., Zhou, C., He, S., Goodyer, P. and Eccles, M. R. (2003). Paired-Box genes are frequently expressed in cancer and often required for cancer cell survival. *Oncogene*, 22, 7989-97. [\[PubMed\]](#)

Relaix, F., Polimeni, M., Rocancourt, D., Ponzetto, C., Schafer, B. W. and Buckingham, M. (2003). The transcriptional activator PAX3-FKHR rescues the defects of Pax3 mutant mice but induces a myogenic gain-of-function phenotype with ligand-independent activation of Met signaling in vivo. *Genes Dev*, 17, 2950-65. [\[PubMed\]](#)

Wollnik, B., Tukel, T., Uyguner, O., Ghanbari, A., Kayserili, H., Emiroglu, M. and Yuksel-Apak, M. (2003). Homozygous and heterozygous inheritance of PAX3 mutations causes different types of Waardenburg syndrome. *Am J Med Genet A*, 122A, 42-5. [\[PubMed\]](#)

Yajima, I., Endo, K., Sato, S., Toyoda, R., Wada, H., Shibahara, S., Numakunai, T., Ikeo, K., Gojobori, T., Goding, C. R., et al. (2003). Cloning and functional analysis of ascidian Mitf in vivo: insights into the origin of vertebrate pigment cells. *Mech Dev*, 120, 1489-504. [\[PubMed\]](#)

Apuzzo, S. and Gros, P. (2002). Cooperative interactions between the two DNA binding domains of Pax3: helix 2 of the paired domain is in the proximity of the amino terminus of the homeodomain. *Biochemistry*, 46, 2984-93. [\[PubMed\]](#)

Barber, T. D., Barber, M. C., Tomescu, O., Barr, F. G., Ruben, S. and Friedman, T. B. (2002). Identification of target genes regulated by PAX3 and PAX3-FKHR in embryogenesis and alveolar rhabdomyosarcoma. *Genomics*, 79, 278-84. [\[PubMed\]](#)

Chi, N. and Epstein, J. A. (2002). Getting your Pax straight: Pax proteins in development and disease. *Trends Genet*, 18, 41-7. [\[PubMed\]](#)

Kamaraju, A. K., Bertolotto, C., Chebath, J. and Revel, M. (2002). Pax3 down-regulation and shut-off of melanogenesis in melanoma B16/F10.9 by interleukin-6 receptor signaling. *J Biol Chem*, 277, 15132-41. [\[PubMed\]](#)

Koushik, S. V., Chen, H., Wang, J. and Conway, S. J. (2002). Generation of a conditional loxP allele of the Pax3 transcription factor that enables selective deletion of the homeodomain. *Genesis*, 32, 114-7. [\[PubMed\]](#)

- Lagutina, I., Conway, S. J., Sublett, J. and Grosveld, G. C. (2002). Pax3-FKHR knock-in mice show developmental aberrations but do not develop tumors. *Mol Cell Biol*, 22, 7204-16. [\[PubMed\]](#)
- Pani, L., Horal, M. and Loeken, M. R. (2002). Rescue of neural tube defects in Pax-3-deficient embryos by p53 loss of function: implications for Pax-3-dependent development and tumorigenesis. *Genes Dev*, 16, 676-80. [\[PubMed\]](#)
- Wiggan, O., Fadel, M. P. and Hamel, P. A. (2002). Pax3 induces cell aggregation and regulates phenotypic mesenchymal-epithelial interconversion. *J Cell Sci*, 115, 517-29. [\[PubMed\]](#)
- Wiggan, O. and Hamel, P. A. (2002). Pax3 regulates morphogenetic cell behavior in vitro coincident with activation of a PCP/non-canonical Wnt-signaling cascade. *J Cell Sci*, 115, 531-41. [\[PubMed\]](#)
- Hornyak, T. J., Hayes, D. J., Chiu, L. Y. and Ziff, E. B. (2001). Transcription factors in melanocyte development: distinct roles for Pax-3 and Mitf. *Mech Dev*, 101, 47-59. [\[PubMed\]](#)
- Hornyak, T. J., Hayes, D. J., Chiu, L. Y. and Ziff, E. B. (2001). Transcription factors in melanocyte development: distinct roles for Pax-3 and Mitf. *Mech Dev*, 101, 47-59. [\[PubMed\]](#)
- Mansouri, A., Pla, P., Larue, L. and Gruss, P. (2001). Pax3 acts cell autonomously in the neural tube and somites by controlling cell surface properties. *Development*, 128, 1995-2005. [\[PubMed\]](#)
- Mayanil, C. S., George, D., Freilich, L., Miljan, E. J., Mania-Farnell, B., Mcclone, D. G. and Bremer, E. G. (2001). Microarray analysis detects novel Pax3 downstream target genes. *J Biol Chem*, 276, 49299-309. [\[PubMed\]](#)
- Scholl, F. A., Kamarashev, J., Murmann, O. V., Geertsen, R., Dummer, R. and Schafer, B. W. (2001). PAX3 is expressed in human melanomas and contributes to tumor cell survival. *Cancer Res*, 61, 823-6. [\[PubMed\]](#)
- Tekin, M., Bodurtha, J. N., Nance, W. E. and Pandya, A. (2001). Waardenburg syndrome type 3 (Klein-Waardenburg syndrome) segregating with a heterozygous deletion in the paired box domain of PAX3: a simple variant or a true syndrome? *Clin Genet*, 60, 301-4. [\[PubMed\]](#)
- Bondurand, N., Pingault, V., Goerlich, D. E., Lemort, N., Sock, E., Le Caignec, C., Wegner, M. and Goossens, M. (2000). Interaction among SOX10, PAX3 and MITF, three genes altered in Waardenburg syndrome. *Hum Mol Genet*, 9, 1907-17. [\[PubMed\]](#)
- Cao, Y. and Wang, C. (2000). The COOH-terminal transactivation domain plays a key role in regulating the in vitro and in vivo function of Pax3 homeodomain. *J Biol Chem*, 275, 9854-62. [\[PubMed\]](#)
- Lang, D., Chen, F., Milewski, R., Li, J., Lu, M. M. and Epstein, J. A. (2000). Pax3 is required for enteric ganglia formation and functions with Sox10 to modulate expression of c-ret. *J Clin Invest*, 106, 963-71. [\[PubMed\]](#)
- Lee, M., Goodall, J., Verastegui, C., Ballotti, R. and Goding, C. R. (2000). Direct regulation of the Microphthalmia promoter by Sox10 links Waardenburg-Shah syndrome (WS4)-associated hypopigmentation and deafness to WS2. *J Biol Chem*, 275, 37978-83. [\[PubMed\]](#)
- Li, J., Chen, F. and Epstein, J. A. (2000). Neural crest expression of Cre recombinase directed by the proximal Pax3 promoter in transgenic mice. *Genesis*, 26, 162-4. [\[PubMed\]](#)
- Margue, C. M., Bernasconi, M., Barr, F. G. and Schafer, B. W. (2000). Transcriptional modulation of the anti-apoptotic protein BCL-XL by the paired box transcription factors PAX3 and PAX3/FKHR. *Oncogene*,

19, 2921-9. [PubMed]

Potterf, S. B., Furumura, M., Dunn, K. J., Arnheiter, H. and Pavan, W. J. (2000). Transcription factor hierarchy in Waardenburg syndrome: regulation of MITF expression by SOX10 and PAX3. *Hum Genet*, 107, 1-6. [PubMed]

Smit, D. J., Smith, A. G., Parsons, P. G., Muscat, G. E. and Sturm, R. A. (2000). Domains of Brn-2 that mediate homodimerization and interaction with general and melanocytic transcription factors. *Eur J Biochem*, 267, 6413-22. [PubMed]

Verastegui, C., Bille, K., Ortonne, J. P. and Ballotti, R. (2000). Regulation of the microphthalmia-associated transcription factor gene by the Waardenburg syndrome type 4 gene, SOX10. *J Biol Chem*, 275, 30757-60. [PubMed]

Barber, T. D., Barber, M. C., Cloutier, T. E. and Friedman, T. B. (1999). PAX3 gene structure, alternative splicing and evolution. *Gene*, 237, 311-9. [PubMed]

Barr, F. G., Fitzgerald, J. C., Ginsberg, J. P., Vanella, M. L., Davis, R. J. and Bennicelli, J. L. (1999). Predominant expression of alternative PAX3 and PAX7 forms in myogenic and neural tumor cell lines. *Cancer Res*, 59, 5443-8. [PubMed]

Fine, E. L., Horal, M., Chang, T. I., Fortin, G. and Loeken, M. R. (1999). Evidence that elevated glucose causes altered gene expression, apoptosis, and neural tube defects in a mouse model of diabetic pregnancy. *Diabetes*, 48, 2454-62. [PubMed]

Galibert, M. D., Yavuzer, U., Dexter, T. J. and Goding, C. R. (1999). Pax3 and regulation of the melanocyte-specific tyrosinase-related protein-1 promoter. *J Biol Chem*, 274, 26894-900. [PubMed]

Hollenbach, A. D., Sublett, J. E., Mcpherson, C. J. and Grosveld, G. (1999). The Pax3-FKHR oncoprotein is unresponsive to the Pax3-associated repressor hDaxx. *EMBO J*, 18, 3702-11. [PubMed]

Li, J., Liu, K. C., Jin, F., Lu, M. M. and Epstein, J. A. (1999). Transgenic rescue of congenital heart disease and spina bifida in Splotch mice. *Development*, 126, 2495-503. [PubMed]

Vachtenheim, J. and Novotna, H. (1999). Expression of genes for microphthalmia isoforms, Pax3 and MSG1, in human melanomas. *Cell Mol Biol (Noisy-le-grand)*, 45, 1075-82. [PubMed]

Hill, A. L., Phelan, S. A. and Loeken, M. R. (1998). Reduced expression of pax-3 is associated with overexpression of cdc46 in the mouse embryo. *Dev Genes Evol*, 208, 128-34. [PubMed]

Magnaghi, P., Roberts, C., Lorain, S., Lipinski, M. and Scambler, P. J. (1998). HIRA, a mammalian homologue of *Saccharomyces cerevisiae* transcriptional co-repressors, interacts with Pax3. *Nat Genet*, 20, 74-7. [PubMed]

Watanabe, A., Takeda, K., Ploplis, B. and Tachibana, M. (1998). Epistatic relationship between Waardenburg syndrome genes MITF and PAX3. *Nat Genet*, 18, 283-6. [PubMed]

Wiggan, O., Taniguchi-Sidle, A. and Hamel, P. A. (1998). Interaction of the pRB-family proteins with factors containing paired-like homeodomains. *Oncogene*, 16, 227-36. [PubMed]

Fortin, A. S., Underhill, D. A. and Gros, P. (1997). Reciprocal effect of Waardenburg syndrome mutations on DNA binding by the Pax-3 paired domain and homeodomain. *Hum Mol Genet*, 6, 1781-90. [PubMed]

Henderson, D. J., Ybot-Gonzalez, P. and Copp, A. J. (1997). Over-expression of the chondroitin sulphate

proteoglycan versican is associated with defective neural crest migration in the Pax3 mutant mouse (splotch). *Mech Dev*, 69, 39-51. [\[PubMed\]](#)

Natoli, T. A., Ellsworth, M. K., Wu, C., Gross, K. W. and Pruitt, S. C. (1997). Positive and negative DNA sequence elements are required to establish the pattern of Pax3 expression. *Development*, 124, 617-26. [\[PubMed\]](#)

Phelan, S. A., Ito, M. and Loeken, M. R. (1997). Neural tube defects in embryos of diabetic mice: role of the Pax-3 gene and apoptosis. *Diabetes*, 46, 1189-97. [\[PubMed\]](#)

Read, A. P. and Newton, V. E. (1997). Waardenburg syndrome. *J Med Genet*, 34, 656-65. [\[PubMed\]](#)

Underhill, D. A. and Gros, P. (1997). The paired-domain regulates DNA binding by the homeodomain within the intact Pax-3 protein. *J Biol Chem*, 272, 14175-82. [\[PubMed\]](#)

Asher, J. H., Jr., Harrison, R. W., Morell, R., Carey, M. L. and Friedman, T. B. (1996a). Effects of Pax3 modifier genes on craniofacial morphology, pigmentation, and viability: a murine model of Waardenburg syndrome variation. *Genomics*, 34, 285-98. [\[PubMed\]](#)

Asher, J. H., Jr., Sommer, A., Morell, R. and Friedman, T. B. (1996b). Missense mutation in the paired domain of PAX3 causes craniofacial-deafness-hand syndrome. *Hum Mutat*, 7, 30-5. [\[PubMed\]](#)

Epstein, J. A., Shapiro, D. N., Cheng, J., Lam, P. Y. and Maas, R. L. (1996). Pax3 modulates expression of the c-Met receptor during limb muscle development. *Proc Natl Acad Sci U S A*, 93, 4213-8. [\[PubMed\]](#)

Ayme, S. and Philip, N. (1995). Possible homozygous Waardenburg syndrome in a fetus with exencephaly. *Am J Med Genet*, 59, 263-5. [\[PubMed\]](#)

Tassabehji, M., Newton, V. E., Liu, X. Z., Brady, A., Donnai, D., Krajewska-Walasek, M., Murday, V., Norman, A., Obersztyn, E., Reardon, W., et al. (1995). The mutational spectrum in Waardenburg syndrome. *Hum Mol Genet*, 4, 2131-7. [\[PubMed\]](#)

Torres, M., Gomez-Pardo, E., Dressler, G. R. and Gruss, P. (1995). Pax-2 controls multiple steps of urogenital development. *Development*, 121, 4057-65. [\[PubMed\]](#)

Underhill, D. A., Vogan, K. J. and Gros, P. (1995). Analysis of the mouse Splotch-delayed mutation indicates that the Pax-3 paired domain can influence homeodomain DNA-binding activity. *Proc Natl Acad Sci U S A*, 92, 3692-6. [\[PubMed\]](#)

Zlotogora, J., Lerer, I., Bar-David, S., Ergaz, Z. and Abeliovich, D. (1995). Homozygosity for Waardenburg syndrome. *Am J Hum Genet*, 56, 1173-8. [\[PubMed\]](#)

Cattanach, B. M., Beechey, C. V., Rasberry, C. and Evans, E. P. (1994). Mutations Pax and Pax. *Mouse Genome*, 92, 503-4.

Chalepakis, G., Goulding, M., Read, A., Strachan, T. and Gruss, P. (1994a). Molecular basis of splotch and Waardenburg Pax-3 mutations. *Proc Natl Acad Sci U S A*, 91, 3685-9. [\[PubMed\]](#)

Chalepakis, G., Jones, F. S., Edelman, G. M. and Gruss, P. (1994b). Pax-3 contains domains for transcription activation and transcription inhibition. *Proc Natl Acad Sci U S A*, 91, 12745-9. [\[PubMed\]](#)

Farrer, L. A., Arnos, K. S., Asher, J. H., Jr., Baldwin, C. T., Diehl, S. R., Friedman, T. B., Greenberg, J., Grundfast, K. M., Hoth, C., Lalwani, A. K., et al. (1994). Locus heterogeneity for Waardenburg syndrome is predictive of clinical subtypes. *Am J Hum Genet*, 55, 728-37. [\[PubMed\]](#)

- Rasberry, C. and Cattanach, B. M. (1994). A new mutation at the Ph locus. *Mouse Genome*, 92, 504-5.
- Tassabehji, M., Newton, V. E., Leverton, K., Turnbull, K., Seemanova, E., Kunze, J., Sperling, K., Strachan, T. and Read, A. P. (1994). PAX3 gene structure and mutations: close analogies between Waardenburg syndrome and the Splotch mouse. *Hum Mol Genet*, 3, 1069-74. [\[PubMed\]](#)
- Tsukamoto, K., Nakamura, Y. and Niikawa, N. (1994). Isolation of two isoforms of the PAX3 gene transcripts and their tissue-specific alternative expression in human adult tissues. *Hum Genet*, 93, 270-4. [\[PubMed\]](#)
- Barr, F. G., Galili, N., Holick, J., Biegel, J. A., Rovera, G. and Emanuel, B. S. (1993). Rearrangement of the PAX3 paired box gene in the paediatric solid tumour alveolar rhabdomyosarcoma. *Nat Genet*, 3, 113-7. [\[PubMed\]](#)
- Franz, T. and Kothary, R. (1993). Characterization of the neural crest defect in Splotch (Sp1H) mutant mice using a lacZ transgene. *Brain Res Dev Brain Res*, 72, 99-105. [\[PubMed\]](#)
- Galili, N., Davis, R. J., Fredericks, W. J., Mukhopadhyay, S., Rauscher, F. J., 3rd, Emanuel, B. S., Rovera, G. and Barr, F. G. (1993). Fusion of a fork head domain gene to PAX3 in the solid tumour alveolar rhabdomyosarcoma. *Nat Genet*, 5, 230-5. [\[PubMed\]](#)
- Goulding, M. D., Lumsden, A. and Gruss, P. (1993). Signals from the notochord and floor plate regulate the region-specific expression of two Pax genes in the developing spinal cord. *Development*, 117, 1001-16. [\[PubMed\]](#)
- Hoth, C. F., Milunsky, A., Lipsky, N., Sheffer, R., Clarren, S. K. and Baldwin, C. T. (1993). Mutations in the paired domain of the human PAX3 gene cause Klein-Waardenburg syndrome (WS-III) as well as Waardenburg syndrome type I (WS-I). *Am J Hum Genet*, 52, 455-62. [\[PubMed\]](#)
- Shapiro, D. N., Sublett, J. E., Li, B., Downing, J. R. and Naeve, C. W. (1993). Fusion of PAX3 to a member of the forkhead family of transcription factors in human alveolar rhabdomyosarcoma. *Cancer Res*, 53, 5108-12. [\[PubMed\]](#)
- Tassabehji, M., Read, A. P., Newton, V. E., Patton, M., Gruss, P., Harris, R. and Strachan, T. (1993). Mutations in the PAX3 gene causing Waardenburg syndrome type 1 and type 2. *Nat Genet*, 3, 26-30. [\[PubMed\]](#)
- Baldwin, C. T., Hoth, C. F., Amos, J. A., Da-Silva, E. O. and Milunsky, A. (1992). An exonic mutation in the HuP2 paired domain gene causes Waardenburg's syndrome. *Nature*, 355, 637-8. [\[PubMed\]](#)
- Glaser, T., Walton, D. S. and Maas, R. L. (1992). Genomic structure, evolutionary conservation and aniridia mutations in the human PAX6 gene. *Nat Genet*, 2, 232-9. [\[PubMed\]](#)
- Jordan, T., Hanson, I., Zaletayev, D., Hodgson, S., Prosser, J., Seawright, A., Hastie, N. and Van Heyningen, V. (1992). The human PAX6 gene is mutated in two patients with aniridia. *Nat Genet*, 1, 328-32. [\[PubMed\]](#)
- Tassabehji, M., Read, A. P., Newton, V. E., Harris, R., Balling, R., Gruss, P. and Strachan, T. (1992). Waardenburg's syndrome patients have mutations in the human homologue of the Pax-3 paired box gene. *Nature*, 355, 635-6. [\[PubMed\]](#)
- Epstein, D. J., Malo, D., Vekemans, M. and Gros, P. (1991a). Molecular characterization of a deletion encompassing the splotch mutation on mouse chromosome 1. *Genomics*, 10, 89-93. [\[PubMed\]](#)
- Epstein, D. J., Vekemans, M. and Gros, P. (1991b). Splotch (Sp2H), a mutation affecting development of the

mouse neural tube, shows a deletion within the paired homeodomain of Pax-3. *Cell*, 67, 767-74. [PubMed]

Goulding, M. D., Chalepakis, G., Deutsch, U., Erselius, J. R. and Gruss, P. (1991). Pax-3, a novel murine DNA binding protein expressed during early neurogenesis. *EMBO J*, 10, 1135-47. [PubMed]

Hill, R. E., Favor, J., Hogan, B. L., Ton, C. C., Saunders, G. F., Hanson, I. M., Prosser, J., Jordan, T., Hastie, N. D. and Van Heyningen, V. (1991). Mouse small eye results from mutations in a paired-like homeobox-containing gene. *Nature*, 354, 522-5. [PubMed]

Burri, M., Tromvoukis, Y., Bopp, D., Frigerio, G. and Noll, M. (1989). Conservation of the paired domain in metazoans and its structure in three isolated human genes. *EMBO J*, 8, 1183-90. [PubMed]

Balling, R., Deutsch, U. and Gruss, P. (1988). Undulated, a mutation affecting the development of the mouse skeleton, has a point mutation in the paired box of Pax 1. *Cell*, 55, 531-5. [PubMed]

Beechey, C. V. and Searle, A. G. (1986). Mutations at the Sp locus. *Mouse News Lett* 75, 28.

Sommer, A., Young-Wee, T. and Frye, T. (1983). Previously undescribed syndrome of craniofacial, hand anomalies, and sensorineural deafness. *Am J Med Genet*, 15, 71-7. [PubMed]

Dickie, M. M. (1964). New Splotch Alleles in the Mouse. *J Hered*, 55, 97-101. [PubMed]

Auerbach, R. (1954). Analysis of the developmental effects of a lethal mutation in the house mouse. *J. Exp. Zool.*, 127, 305-329.

Russell, W. L. and Roscoe, B. (1947). Splotch, a new mutation in the house mouse. *Genetics*, 32, 102.